

Publikationen von Dr. med. Bertold Schrank:

1. Schrank B., von Pein H, Sommer C, Haas D, Hörtnagel K, Biskup S: Dropped Head Syndrom als Folge einer behandelbaren metabolischen Myopathie. *Klin Neurophysiol* 2017; 48(02): 102-106.
2. Schrank B, Schoser B, Klopstock T, Schneiderat P, Horvath R, Abicht A, Holinski-Feder E, Augustis S: Lifetime exercise intolerance with lactic acidosis as key manifestation of novel compound heterozygous ACAD9 mutations causing complex I deficiency. *Neuromuscul Disord.* 2017 27(5):473-476.
3. Werling S, Schrank B, Eckardt AJ, Hauburger A, Deschauer M, Müller M.: Oculopharyngeal muscular dystrophy as a rare cause of dysphagia. *Ann Gastroenterol.* 2015 Apr-Jun;28(2):291-293.
4. Dorst J, Dupuis L, Petri S, Kollewe K, Abdulla S, Wolf J, Weber M, Czell D, Burkhardt C, Hanisch F, Vielhaber S, Meyer T, Frisch G, Kettemann D, Grehl T, Schrank B, Ludolph AC. Percutaneous endoscopic gastrostomy in amyotrophic lateral sclerosis: a prospective observational study. *J Neurol.* 2015 Apr; 262(4):849-58.
5. Schrank B: M. Pompe. Diagnostik und Therapie der Glykogenose Typ II. *Thieme Praxis Report* 2015; 7 (11): 3-15.
6. Schara U, Schneider-Gold C, Schrank B: Klinik und Transition neuromuskulärer Erkrankungen . Springer, Heidelberg, 2015
7. Schrank B: CME Morbus Pompe/ Glykogenose Typ II. Georg Thieme Verlag Stuttgart, 2013 (4. Aufl.)
8. Sieb J.P., Schrank B.: Neuromuskuläre Erkrankungen Kohlhammer Verlag Stuttgart, 2009, 1. Auflage
9. Walter MC, Reilich P, Thiele S, Schessl J, Schreiber H, Reiners K, Kress W, Müller-Reible C, Vorgerd M, Urban P, Schrank B, Deschauer M, Schlotter-Weigel B, Kohnen R, Lochmüller H. Treatment of dysferlinopathy with deflazacort: a double-blind, placebo-controlled clinical trial. *Orphanet J Rare Dis.* 2013 Feb 14;8:26.
10. Sarkozy A, Deschauer M, Carlier RY, Schrank B, Seeger J, Walter MC, Schoser B, Reilich P, Leturq F, Radunovic A, Behin A, Laforet P, Eymard B, Schreiber H, Hicks D, Vaidya SS, Gläser D, Carlier PG, Bushby K, Lochmüller H, Straub V.: Muscle MRI findings in limb girdle muscular dystrophy type 2L. *Neuromuscul Disord.* 2012 Oct 1;22 Suppl 2:S122-9.
11. Dupuis L, Dengler R, Heneka MT, Meyer T, Zierz S, Kassubek J, Fischer W, Steiner F, Lindauer E, Otto M, Dreyhaupt J, Grehl T, Hermann A, Winkler AS, Bogdahn U, Benecke R, Schrank B, Wessig C, Grosskreutz J, Ludolph AC; GERP ALS Study Group. A randomized, double blind, placebo-controlled trial of pioglitazone in combination with riluzole in amyotrophic lateral sclerosis. *PLoS One.* 2012;7(6):e37885.

12. Seeger J, Schrank B, Pyle A, Stucka R, Lörcher U, Müller-Ziermann S, Abicht A, Czermin B, Holinski-Feder E, Lochmüller H, Horvath R.: Clinical and neuropathological findings in patients with TACO1 mutations. *Neuromuscul Disord.* 2010 Nov;20(11):720-4.
13. Schrank B: Atemmuskelschwäche: Differentialdiagnose – klinisch und elektrophysiologisch. *Das Neurophysiologie-Labor*, Volume 32, Issue 3, September 2010, Pages 163-169
14. Winter Y, Schepelmann K, Spottke AE, Claus D, Grothe C, Schröder R, Heuss D, Vielhaber S, Tackenberg B, Mylius V, Reese JP, Kiefer R, Schrank B, Oertel WH, Dodel R.: Health-related quality of life in ALS, myasthenia gravis and facioscapulohumeral muscular dystrophy. *J Neurol.* 2010 Sep;257(9):1473-81.
15. Wirth B, Brichta L, Schrank B, Lochmüller H, Blick S, Baasner A, Heller R. Mildly affected patients with spinal muscular atrophy are partially protected by an increased SMN2 copy number. *Hum Genet.* 2006 May;119(4):422-8.
16. B. Schrank, P. Urban, U. Lörcher: Der Einsatz der Magnetresonanztomographie der Muskulatur bei der Diagnose neuromuskulärer Erkrankungen. *Klin Neuroradiol* 2005; 15: 241-255
17. Graf M, Ecker D, Horowski R et al.: High dose vitamin E therapy in amyotrophic lateral sclerosis as add-on therapy to riluzole: results of a placebo-controlled double-blind study. *J Neural Transm.* 2005;112(5): 649-60.
18. Zeiler B, Oelze R, Schrank B: ALS-Kachexie: Gewichtsverlust ist ein Frühzeichen motorischer Systemerkrankungen mit trunkalem Beginn. *Aktuelle Neurologie* 2005; 32: S260
19. Schrank B, Meng G et al. (2002): Adult-Onset Spinal Muscular Atrophy due to Loss of the SMN1 Gene Manifesting as Segmental Anterior Horn Cell Disease. *Neurology*, 58, Suppl.3, A465
20. Sieb JP, Kraner S, Schrank B, Reitter B, Goebel TH, Tzartos SJ, Steinlein OK: Severe congenital myasthenic syndrome due to homozygosity of the 1293insG epsilon-acetylcholine receptor subunit mutation. *Ann Neurol.* 2000 Sep;48(3):379-83.
21. Jablonka S, Rossoll W, Schrank B, Sendtner M: The role of SMN in spinal muscular atrophy. *J Neurol.* 2000 Mar;247 Suppl 1:137-42.
22. Jablonka S, Schrank B, Kralewski M, Rossoll W, Sendtner M. Reduced survival motor neuron (Smn) gene dose in mice leads to motor neuron degeneration: an animal model for spinal muscular atrophy type III. *Hum Mol Genet.* 2000 Feb 12;9(3):341-6.
23. Monani UR, Sendtner M, Covert DD, Parsons DW, Andreassi C, Le TT, Jablonka S, Schrank B, Rossoll W, Prior TW, Morris GE, Burghes AH. The human centromeric survival motor neuron gene (SMN2) rescues embryonic lethality in Smn(-/-) mice and results in a mouse with spinal muscular atrophy. *Hum Mol Genet.* 2000 Feb 12;9(3):333-9.

24. Giess R, Goetz R, Schrank B, Ochs G, Sendtner M, Toyka K. Potential implications of a ciliary neurotrophic factor gene mutation in a German population of patients with motor neuron disease. *Muscle Nerve*. 1998 Feb;21(2):236-8.
25. Schrank B, Gotz R, Gunnarsen JM, Ure JM, Toyka KV, Smith AG, Sendtner M.: Inactivation of the survival motor neuron gene, a candidate gene for human spinal muscular atrophy, leads to massive cell death in early mouse embryos. *Proc Natl Acad Sci U S A*. 1997 Sep 2;94(18):9920-5.
26. Winterholler M, Claus D, Bockelbrink A, Borasio GD, Pongratz D, Schrank B, Toyka KV, Neundorfer B. [Recommendations of Bavarian Muscle Centers of the German Neuromuscular Disease Society for home ventilation of neuromuscular diseases of adult patients] *Nervenarzt*. 1997 Apr;68(4):351-7. Review.
27. Liu J, Schrank B, Waterston RH. Interaction between a putative mechanosensory membrane channel and a collagen. *Science*. 1996 Jul 19; 273(5273):361-4.
28. Petersen RB, Tabaton M, Berg L, Schrank B, Torack RM, Leal S, Julien J, Vital C, Deleplanque B, Pendlebury WW, et al. Analysis of the prion protein gene in thalamic dementia. *Neurology*. 1992 Oct;42(10):1859-63.
29. Goldfarb LG, Petersen RB, Tabaton M, Brown P, LeBlanc AC, Montagna P, Cortelli P, Julien J, Vital C, Pendlebury WW, et al. Fatal familial insomnia and familial Creutzfeldt-Jakob disease: disease phenotype determined by a DNA polymorphism. *Science*. 1992 Oct 30; 258(5083):806-8.
30. Williams BD, Schrank B, Huynh C, Shownkeen R, Waterston RH. A genetic mapping system in *Caenorhabditis elegans* based on polymorphic sequence-tagged sites. *Genetics*. 1992 Jul; 131(3):609-24.